<110> Samsung Electronics Co. Ltd

<120> A variant of HNF-1a gene having novel single nucleotide polymorphism and a variant protein encoded by the same

<130> YPL-0064

<140> 10/6663,857
<141> 2003-09-15

<160> 27

<170> KopatentIn 1.71

<210> 1
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<212> DNA
<213> Homo sapiens

<220>
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<223> amino acid sequence of HNF-1a

<400> 1

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Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu
  1               5                  10                  15

ctc gag tca ggg ctg agc aaa gag gca ctg atc cag gca ctg ggt gag          96
Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu
             20                  25                  30

ccg ggg ccc tac ctc ctg gct gga gaa ggc ccc ctg gac aag ggg gag         144
Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu
         35                  40                  45

tcc tgc ggc ggc ggt cga ggg gag ctg gct gag ctg ccc aat ggg ctg         192
Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu
     50                  55                  60

ggg gag act cgg ggc tcc gag gac gag acg gac gac gat ggg gaa gac         240
Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp
 65                  70                  75                  80

ttc acg cca ccc atc ctc aaa gag ctg gag aac ctc agc cct gag gag         288
Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu
                 85                  90                  95

gcg gcc cac cag aaa gcc gtg gtg gag acc ctt ctg cag gag gac ccg         336
Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro
            100                 105                 110

tgg cgt gtg gcg aag atg gtc aag tcc tac ctg cag cag cac aac atc         384
Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile
        115                 120                 125

cca cag cgg gag gtg gtc gat acc act ggc ctc aac cag tcc cac ctg         432
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Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu
    130                 135                 140

tcc caa cac ctc aac aag ggc act ccc atg aag acg cag aag cgg gcc          480
Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala
145                 150                 155                 160

gcc ctg tac acc tgg tac gtc cgc aag cag cga gag gtg gcg cag cag          528
Ala Leu Tyr Thr Trp Tyr Val Arg Lys Gln Arg Glu Val Ala Gln Gln
                165                 170                 175

ttc acc cat gca ggg cag gga ggg ctg att gaa gag ccc aca ggt gat          576
Phe Thr His Ala Gly Gln Gly Gly Leu Ile Glu Glu Pro Thr Gly Asp
            180                 185                 190

gag cta cca acc aag aag ggg cgg agg aac cgt ttc aag tgg ggc cca          624
Glu Leu Pro Thr Lys Lys Gly Arg Arg Asn Arg Phe Lys Trp Gly Pro
        195                 200                 205

gca tcc cag cag atc ctg ttc cag gcc tat gag agg cag aag aac cct          672
Ala Ser Gln Gln Ile Leu Phe Gln Ala Tyr Glu Arg Gln Lys Asn Pro
    210                 215                 220

agc aag gag gag cga gag acg cta gtg gag gag tgc aat agg gcg gaa          720
Ser Lys Glu Glu Arg Glu Thr Leu Val Glu Glu Cys Asn Arg Ala Glu
225                 230                 235                 240

tgc atc cag aga ggg gtg tcc cca tca cag gca cag ggg ctg ggc tcc          768
Cys Ile Gln Arg Gly Val Ser Pro Ser Gln Ala Gln Gly Leu Gly Ser
                245                 250                 255

aac ctc gtc acg gag gtg cgt gtc tac aac tgg ttt gcc aac cgg cgc          816
Asn Leu Val Thr Glu Val Arg Val Tyr Asn Trp Phe Ala Asn Arg Arg
            260                 265                 270

aaa gaa gaa gcc ttc cgg cac aag ctg gcc atg gac acg tac agc ggg          864
Lys Glu Glu Ala Phe Arg His Lys Leu Ala Met Asp Thr Tyr Ser Gly
        275                 280                 285

ccc ccc cca ggg cca ggc ccg gga cct gcg ctg ccc gct cac agc tcc          912
Pro Pro Pro Gly Pro Gly Pro Gly Pro Ala Leu Pro Ala His Ser Ser
    290                 295                 300

cct ggc ctg cct cca cct gcc ctc tcc ccc agt aag gtc cac ggt gtg          960
Pro Gly Leu Pro Pro Pro Ala Leu Ser Pro Ser Lys Val His Gly Val
305                 310                 315                 320

cgc tat gga cag cct gcg acc agt gag act gca gaa gta ccc tca agc         1008
Arg Tyr Gly Gln Pro Ala Thr Ser Glu Thr Ala Glu Val Pro Ser Ser
                325                 330                 335

agc ggc ggt ccc tta gtg aca gtg tct aca ccc ctc cac caa gtg tcc         1056
Ser Gly Gly Pro Leu Val Thr Val Ser Thr Pro Leu His Gln Val Ser
            340                 345                 350

ccc acg ggc ctg gag ccc agc cac agc ctg ctg agt aca gaa gcc aag         1104
Pro Thr Gly Leu Glu Pro Ser His Ser Leu Leu Ser Thr Glu Ala Lys
        355                 360                 365
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ctg gtc tca gca gct ggg ggc ccc ctc ccc cct gtc agc acc ctg aca        1152
Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr
    370                 375                 380

gca ctg cac agc ttg gag cag aca tcc cca ggc ctc aac cag cag ccc        1200
Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro
385                 390                 395                 400

cag aac ctc atc atg gcc tca ctt cct ggg gtc atg acc atc ggg cct        1248
Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro
                405                 410                 415

ggt gag cct gcc tcc ctg ggt cct acg ttc acc aac aca ggt gcc tcc        1296
Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser
            420                 425                 430

acc ctg gtc atc ggc ctg gcc tcc acg cag gca cag agt gtg ccg gtc        1344
Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val
        435                 440                 445

atc aac agc atg ggc agc agc ctg acc acc ctg cag ccc gtc cag ttc        1392
Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe
    450                 455                 460

tcc cag ccg ctg cac ccc tcc tac cag cag ccg ctc atg cca cct gtg        1440
Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val
465                 470                 475                 480

cag agc cat gtg acc cag aac ccc ttc atg gcc acc atg gct cag ctg        1488
Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu
                485                 490                 495

cag agc ccc cac gcc ctc tac agc cac aag ccc gag gtg gcc cag tac        1536
Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr
            500                 505                 510

acc cac acg ggc ctg ctc ccg cag act atg ctc atc acc gac acc acc        1584
Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr
        515                 520                 525

aac ctg agc gcc ctg gcc agc ctc acg ccc acc aag cag gtc ttc acc        1632
Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr
    530                 535                 540

tca gac act gag gcc tcc agt gag tcc ggg ctt cac acg ccg gca tct        1680
Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser
545                 550                 555                 560

cag gcc acc acc ctc cac atc ccc agc cag gac cct gcc ggc atc cag        1728
Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln
                565                 570                 575

cac ctg cag ccg gcc cac cgg ctc agc gcc agc ccc aca gtg tcc tcc        1776
His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser
            580                 585                 590

agc agc ctg gtg ctg tac cag agc tca gac tcc agc aat ggc cag agc        1824
Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser
        595                 600                 605
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cac ctg ctg cca tcc aac cac agc gtc atc gag acc ttc atc tcc acc          1872
His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr
    610                 615                 620

cag atg gcc tct tcc tcc cag         taa                              1896
Gln Met Ala Ser Ser Ser Gln
625                 630
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<212> PRT
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<400> 2

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Met Val Ser Lys Leu Ser Gln Leu Gln Thr Glu Leu Leu Ala Ala Leu
  1               5                  10                  15

Leu Glu Ser Gly Leu Ser Lys Glu Ala Leu Ile Gln Ala Leu Gly Glu
             20                  25                  30

Pro Gly Pro Tyr Leu Leu Ala Gly Glu Gly Pro Leu Asp Lys Gly Glu
         35                  40                  45

Ser Cys Gly Gly Gly Arg Gly Glu Leu Ala Glu Leu Pro Asn Gly Leu
     50                  55                  60

Gly Glu Thr Arg Gly Ser Glu Asp Glu Thr Asp Asp Asp Gly Glu Asp
 65                  70                  75                  80

Phe Thr Pro Pro Ile Leu Lys Glu Leu Glu Asn Leu Ser Pro Glu Glu
                 85                  90                  95

Ala Ala His Gln Lys Ala Val Val Glu Thr Leu Leu Gln Glu Asp Pro
            100                 105                 110

Trp Arg Val Ala Lys Met Val Lys Ser Tyr Leu Gln Gln His Asn Ile
        115                 120                 125

Pro Gln Arg Glu Val Val Asp Thr Thr Gly Leu Asn Gln Ser His Leu
    130                 135                 140

Ser Gln His Leu Asn Lys Gly Thr Pro Met Lys Thr Gln Lys Arg Ala
145                 150                 155                 160

Ala Leu Tyr Thr Trp Tyr Val Arg Lys Gln Arg Glu Val Ala Gln Gln
                165                 170                 175

Phe Thr His Ala Gly Gln Gly Gly Leu Ile Glu Glu Pro Thr Gly Asp
            180                 185                 190

Glu Leu Pro Thr Lys Lys Gly Arg Arg Asn Arg Phe Lys Trp Gly Pro
        195                 200                 205

Ala Ser Gln Gln Ile Leu Phe Gln Ala Tyr Glu Arg Gln Lys Asn Pro
    210                 215                 220

Ser Lys Glu Glu Arg Glu Thr Leu Val Glu Glu Cys Asn Arg Ala Glu
```

225 230 235 240

Cys Ile Gln Arg Gly Val Ser Pro Ser Gln Ala Gln Gly Leu Gly Ser
245 250 255

Asn Leu Val Thr Glu Val Arg Val Tyr Asn Trp Phe Ala Asn Arg Arg
260 265 270

Lys Glu Glu Ala Phe Arg His Lys Leu Ala Met Asp Thr Tyr Ser Gly
275 280 285

Pro Pro Pro Gly Pro Gly Pro Gly Pro Ala Leu Pro Ala His Ser Ser
290 295 300

Pro Gly Leu Pro Pro Pro Ala Leu Ser Pro Ser Lys Val His Gly Val
305 310 315 320

Arg Tyr Gly Gln Pro Ala Thr Ser Glu Thr Ala Glu Val Pro Ser Ser
325 330 335

Ser Gly Gly Pro Leu Val Thr Val Ser Thr Pro Leu His Gln Val Ser
340 345 350

Pro Thr Gly Leu Glu Pro Ser His Ser Leu Leu Ser Thr Glu Ala Lys
355 360 365

Leu Val Ser Ala Ala Gly Gly Pro Leu Pro Pro Val Ser Thr Leu Thr
370 375 380

Ala Leu His Ser Leu Glu Gln Thr Ser Pro Gly Leu Asn Gln Gln Pro
385 390 395 400

Gln Asn Leu Ile Met Ala Ser Leu Pro Gly Val Met Thr Ile Gly Pro
405 410 415

Gly Glu Pro Ala Ser Leu Gly Pro Thr Phe Thr Asn Thr Gly Ala Ser
420 425 430

Thr Leu Val Ile Gly Leu Ala Ser Thr Gln Ala Gln Ser Val Pro Val
435 440 445

Ile Asn Ser Met Gly Ser Ser Leu Thr Thr Leu Gln Pro Val Gln Phe
450 455 460

Ser Gln Pro Leu His Pro Ser Tyr Gln Gln Pro Leu Met Pro Pro Val
465 470 475 480

Gln Ser His Val Thr Gln Asn Pro Phe Met Ala Thr Met Ala Gln Leu
485 490 495

Gln Ser Pro His Ala Leu Tyr Ser His Lys Pro Glu Val Ala Gln Tyr
500 505 510

Thr His Thr Gly Leu Leu Pro Gln Thr Met Leu Ile Thr Asp Thr Thr
515 520 525

Asn Leu Ser Ala Leu Ala Ser Leu Thr Pro Thr Lys Gln Val Phe Thr
530 535 540

```
Ser Asp Thr Glu Ala Ser Ser Glu Ser Gly Leu His Thr Pro Ala Ser
545                 550                 555                 560

Gln Ala Thr Thr Leu His Ile Pro Ser Gln Asp Pro Ala Gly Ile Gln
                565                 570                 575

His Leu Gln Pro Ala His Arg Leu Ser Ala Ser Pro Thr Val Ser Ser
            580                 585                 590

Ser Ser Leu Val Leu Tyr Gln Ser Ser Asp Ser Ser Asn Gly Gln Ser
        595                 600                 605

His Leu Leu Pro Ser Asn His Ser Val Ile Glu Thr Phe Ile Ser Thr
    610                 615                 620

Gln Met Ala Ser Ser Ser Gln
625                 630


<210>    3
<211>    93
<212>    DNA
<213>    Homo sapiens

<400>    3
gtaaggtcca ggcctgctgg ccctcccttg gcctgtgaca gagcccctca cccccacatc          60

ccccgggctc aggaggctgc tctgctcccc cag                                       93


<210>    4
<211>    41
<212>    DNA
<213>    Artificial Sequence

<220>
<223>    sense primer for amplifying promoter of MODY3 gene


<400>    4
taatacgact cactataggg tggccgtgag catcctctgc c                              41


<210>    5
<211>    39
<212>    DNA
<213>    Artificial Sequence

<220>
<223>    antisense primer for amplifying promoter of MODY3 gene


<400>    5
gtaaccctca ctaaagggac gtgggttgcg tttgcctgc                                 39


<210>    6
<211>    40
<212>    DNA
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<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 1 of MODY3 gene

<400> 6
taatacgact cactataggg cgtggccctg tggcagccga 40

<210> 7
<211> 40
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying exon 1 of MODY3 gene

<400> 7
gtaaccctca ctaaagggag ggctcgttag gagctgaggg 40

<210> 8
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<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 2 of MODY3 gene

<400> 8
taatacgact cactataggg cccttgctga gcagatcccg tc 42

<210> 9
<211> 40
<212> DNA
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<220>
<223> antisense primer for amplifying exon 2 of MODY3 gene

<400> 9
gtaaccctca ctaaagggag ggatggtgaa gcttccagcc 40

<210> 10
<211> 40
<212> DNA
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<220>
<223> sense primer for amplifying exon 3 of MODY3 gene

<400> 10

taatacgact cactataggg gcaaggtcag gggaatggac 40

<210> 11
<211> 42
<212> DNA
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<220>
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<400> 11
gtaaccctca ctaaagggac gccgttgtac ctattgcact cc 42

<210> 12
<211> 43
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 4 of MODY3 gene

<400> 12
taatacgact cactataggg ggctcatggg tggctatttc tgc 43

<210> 13
<211> 42
<212> DNA
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<220>
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<400> 13
gtaaccctca ctaaagggac gtgtcccttg tccccacata cc 42

<210> 14
<211> 42
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 5 of MODY3 gene

<400> 14
taatacgact cactataggg tgctgaggca ggacactgct tc 42

<210> 15
<211> 42
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying exon 5 of MODY3 gene

<400> 15
gtaaccctca ctaaagggat acaagcaagg acactcacca gc 42

<210> 16
<211> 41
<212> DNA
<213> Artificial Sequence

<220>
<223> sense primer for amplifying exon 6 of MODY3 gene

<400> 16
taatacgact cactataggg cccggacaca gcttggcttc c 41

<210> 17
<211> 42
<212> DNA
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<220>
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<400> 17
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<210> 18
<211> 40
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<220>
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<400> 18
taatacgact cactataggg caggcctggc ctccacgcag 40

<210> 19
<211> 40
<212> DNA
<213> Artificial Sequence

<220>
<223> antisense primer for amplifying exon 7 of MODY3 gene

<400> 19
gtaaccctca ctaaagggag gggctctgca gctgagccat 40

<210> 20
<211> 41
<212> DNA
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<220>
<223> sense primer for amplifying exon 8 and 9 of MODY3 gene

<400> 20
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<210> 21
<211> 40
<212> DNA
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<220>
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<400> 21
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<210> 22
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<212> DNA
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<220>
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<400> 22
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<210> 23
<211> 41
<212> DNA
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<220>
<223> antisense primer for amplifying exon 10 of MODY3 gene

<400> 23
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<210> 24
<211> 20
<212> DNA
<213> Artificial Sequence

<220>
<223> T7 promoter sequence

<400> 24
taatacgact cactataggg 20

<210> 25
<211> 19
<212> DNA
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<220>
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<400> 25
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<210> 26
<211> 540
<212> DNA
<213> Homo sapiens

<400> 26
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ccagccctct acagccacaa gcccgaggtg gcccagtaca cccacacggg cctgctcccg 120

cagactatgc tcatcaccga caccaccaac ctgagcgccc tggccagcct cacgcccacc 180

aagcaggtaa ggtccaggcc tgctggccct ccctcggcct gtgacagagc ccctcacccc 240

cacatccccc gggctcagga ggctgctctg ctcccccagg tcttcacctc agacactgag 300

gcctccagtg agtccgggct tcacacgccg gcatctcagg ccaccaccct ccacatcccc 360

agccaggacc ctgccggcat ccagcacctg cagccggccc accggctcag cgccagcccc 420

acaggtgaga ggccctggct ccacccccctc ccttactgtc cctgcccccct tccatgttgg 480

tcccacccct tctgttgctg tccgtcactg tggggctgtg catgcagcag gcctagggct 540

540

<210> 27
<211> 540
<212> DNA
<213> Homo sapiens

<400> 27
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ccagccctct acagccacaa gcccgaggtg gcccagtaca cccacacggg cctgctcccg 120

cagactatgc tcatcaccga caccaccaac ctgagcgccc tggccagcct cacgcccacc 180

aagcaggtaa ggtccaggcc tgctggccct cccttggcct gtgacagagc ccctcacccc 240

cacatccccc gggctcagga ggctgctctg ctcccccagg tcttcacctc agacactgag 300

gcctccagtg agtccgggct tcacacgccg gcatctcagg ccaccaccct ccacgtcccc 360

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acaggtgaga ggccctggct ccacccccctc ccttactgtc cctgcccccct tccatgttgg 480

tcccaccccct tctgttgctg tccgtcactg tggggctgtg catgcagcag gcctagggct 540

540